To Print: Click your browser's PRINT button.
NOTE: To view the article with Web enhancements, go to:
http://www.medscape.com/viewarticle/583526

REUTERS HEALTH INFORMATION

New Technique Reveals Genetic Copy Number Variations With SIDS

By Anthony J. Brown, MD

NEW YORK (Reuters Health) Nov 13 - US researchers have shown that de novo copy number variations are present in some cases of sudden infant death syndrome (SIDS). Although further research is needed to confirm and expand on the findings, the results point to histone cluster genes as possible players in SIDS.

"Cytogenetic abnormalities in sudden infant death...have been mentioned since the 1970s," lead author Dr. Gokce A. Toruner told Reuters Health. "What is novel about this study is the utilization of a microarray technology, called array-based comparative genome hybridization (array-CGH), for the analysis of the specimens."

This technology, added Dr. Toruner, a researcher with UMDNJ-NJ Medical School in Newark, New Jersey, provides much greater genetic detail than routine karyotypic analysis. "If we use a 'Yahoo Map' analogy, the resolution of karyotyping is like a 'Country Level' map, whereas the resolution of the array-CGH technology...is a 'City Level' map."

The current study, which was presented Thursday at the American Society of Human Genetics annual meeting in Philadelphia, featured array-CGH testing of tissue specimens from 27 infants who died from SIDS and from their immediate family members.

Copy number variations were identified in three (11%) of the cases. An inherited balanced translocation was seen in one case, whereas in the other two the variations arose de novo.

The two cases with de novo copy number variations had an overlapping deletion and duplication on chromosome 6p22, suggesting that this region may play a key role in the etiology of SIDS. This region, Dr. Toruner noted, contains genes that encode histones, which are essential for DNA organization and thought to regulate global gene expression.

Moving forward, Dr. Toruner said that "larger studies with higher resolution arrays are needed. In addition, the candidate genes (histone cluster genes)...must be analyzed with different genetic analysis techniques, since not only copy number changes, but also point mutations in these genes may be important in the disease process."